http://www.ncbi.nlm.nih.gov/omim/

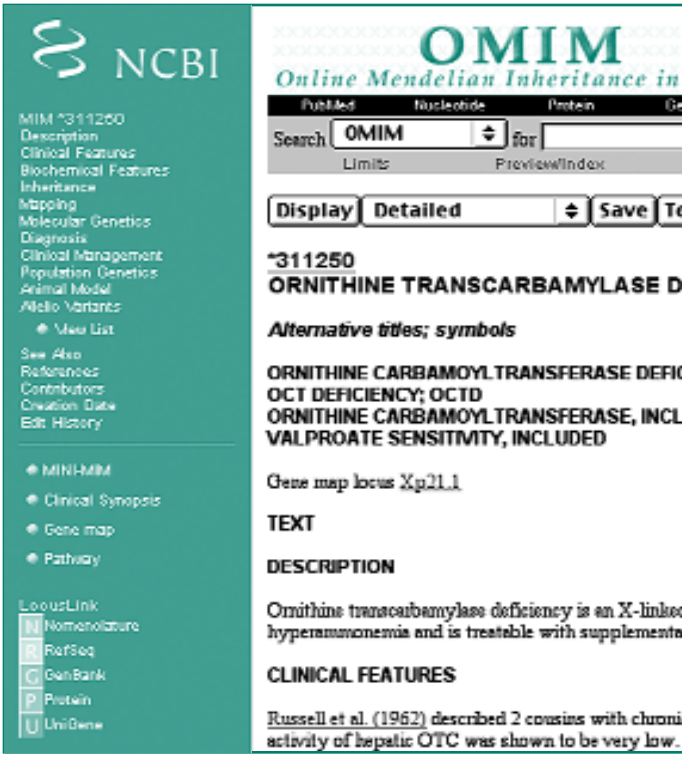
Online *Mendelian Inheritance in Man* (OMIM™) is a continuously updated catalog of human genes and genetic disorders. *Mendelian Inheritance in Man* has been maintained on computer since late 1963, and was a pioneer in computer-based publication with its first book edition in 1966 (and 12th in 1998). See McKusick, V.A.: *Mendelian Inheritance in Man*. Baltimore: Johns Hopkins University Press, 1966, 1968, 1971, 1975, 1978, 1983, 1986, 1988, 1990, 1992, 1994, 1998.

Public access to OMIM is available from the National Center for Biotechnology Information (NCBI) **(http://www.ncbi.nlm.nih.gov/omim/).** OMIM has also recently been integrated into NCBI's Entrez retrieval system, as shown here in the Ornithine Transcarbamylase Deficiency example. Questions about OMIM access may be directed to NCBI via e-mail (info@ncbi.nlm.nih.gov) or telephone (301) 496-2475.

FEATURES

In addition to curated, full-text descriptions of genes and genetic disorders, OMIM has copious links to other databases, including PubMed, DNA and Protein sequences via Entrez, UniGene, LocusLink, Human Genome Map Viewer, the Human Gene Mutation Database (HGMD), Mouse Genome Database (MGD), Online Mendelian Inheritance in Animals (OMIA), and more.

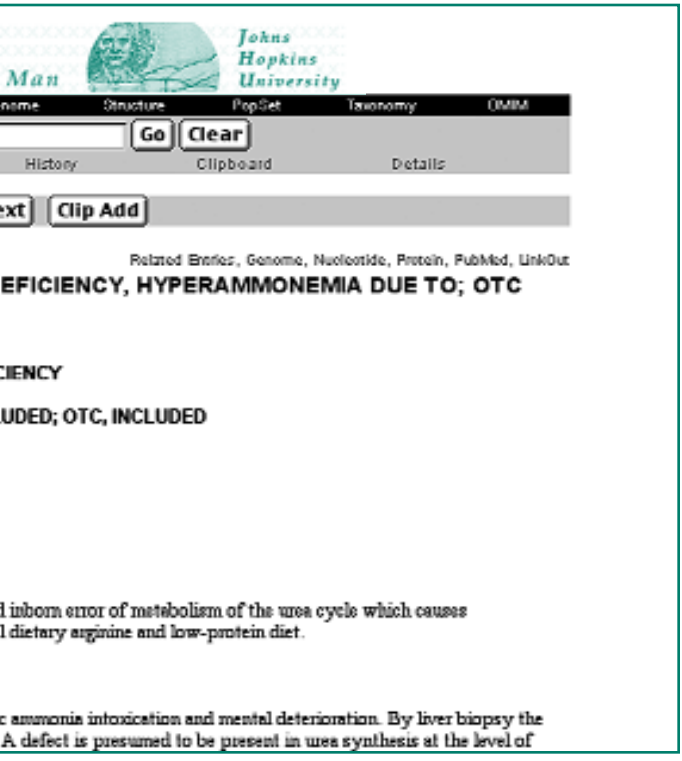
NCBI's powerful neighboring feature has been applied to OMIM. Pressing the lightbulb symbol at the end of a paragraph will retrieve citations in PubMed whose text most closely matches the OMIM text.



References in OMIM are linked to PubMed. Abstracts of journal articles are available by clicking on the PubMed ID after each reference.

78. Oppliger Leibundgut, E.; Liechti-Gallati, S.; Colombo, J.-P.; Wermuth, B.:
Ornithine transcarbamylase deficiency: ten new mutations and high proportion of de novo mutations in heterozygous females.
Hum. Mutat. 9: 409-411, 1997.
PubMed ID: 9143919

OMIM's Synopsis of the Human Gene Map and its clinical subset, the Morbid Map, can be searched independently and are displayed in tabular form. Shortened versions of the longest entries and condensed lists of clinical features for phenotypes are available through the Mini-MIM and Clinical Synopsis links.



CLINICAL SYNOPSIS

(partial)

INHERITANCE:
- X-linked recessive

GROWTH:
- **Other**
  - Failure to thrive

ABDOMEN:
- **Gastrointestinal**
  - Protein avoidance
  - Vomiting

NEUROLOGIC:
- **Central nervous system**
  - Irritability
  - Lethargy
  - Episodic ataxia
  - Coma
  - Seizures
  - Cerebral edema
  - Developmental delay
  - Mental retardation

METABOLIC FEATURES:
- Episodic ammonia intoxication
- Respiratory alkalosis

As of September 25, 2002, OMIM had 13,922 entries and continues to update over 600 entries and add over 100 new entries per month. The Morbid Map lists over 2,940 disorders spread over 2,164 loci. OMIM is a popular resource for clinicians and researchers, registering over 70,000 queries and 8,000 unique users per day. Curation of the database and editorial decisions take place at Johns Hopkins University School of Medicine. Authors are located at Johns Hopkins and around the world.

OMIM

Online Mendelian Inheritance in Man